

Genetic and Rare Diseases Information Center

Keeping pace with the ever-expanding body of scientific knowledge about genetic and rare diseases can be an uphill battle for many health-care professionals. Even for those who manage to stay abreast of the latest developments, a major challenge remains: finding the time and resources needed to clearly convey such information to patients and their families.

Now, the National Institutes of Health (NIH) is offering health-care professionals free assistance on both fronts in the form of its recently established Genetics and Rare Diseases (GARD) Information Center. Funded by the NIH's National Human Genome Research Institute (NHGRI) and the Office of Rare Diseases (ORD), the center provides health-care professionals and their patients with immediate access to experienced information specialists who can furnish current and accurate information about more than 6,000 genetic and rare diseases.

Since it was established in February 2002, GARD has responded to nearly 4,000 inquiries on rare and genetic diseases. The requests include many inquiries from physicians, nurses and other health-care professionals, as well as patients and patients' families directed to the site by health-care professionals.

To date, most users have contacted GARD by e-mail, GARDinfo@nih.gov, or by calling the toll-free telephone: (888) 205-2311 or TTY: (888)205-3223, which are answered from noon to 6 p.m., Monday through Friday. Requests can also be sent to GARD by fax: (240) 632-9164; or by mail: P.O. Box 8126, Gaithersburg, MD 20898-8126.

People requesting information receive a response from GARD within four to seven business days on average. However, depending on the nature of the inquiry, the turnaround time for inquiries from health-care professionals often is much faster. For example, an urgent request may receive a response within 24 hours or even immediately.

However, GARD's success is reflected by far more than statistics. Health-care professionals who have taken advantage of the services offered by the center have responded enthusiastically. In the words of one physician: "Thank you very much for the information, which is very helpful ... the genetic terminology doesn't bother me too much – just takes me back to my student days."

Feedback from health-care professionals plays a critical role in GARD's ongoing effort to fine tune the services that it provides. For example, based on comments from some of its initial users, GARD now offers even more comprehensive responses with greater detail on locating information resources than when the center first opened.

“You have started me in the right direction and helped me greatly as I have not been able to find any information about this disorder to date,” wrote one health-care professional, adding: “I will share this information with my client’s maternal grandmother and foster mother.”

GARD information specialists provide inquirers with current, accurate and authoritative information by drawing from public domain sources, including reliable Web sites, brochures, articles and organizations.

While patients and their families often contact GARD seeking direct referrals to health-care professionals or asking for treatment or medical management suggestions, it is important to emphasize that GARD does not directly provide this type of information. Instead, GARD information specialists direct the inquirer to resources that might give treatment information, e.g. journal articles or clinical trials. GARD also does not provide genetic counseling or offer diagnostic testing, but will point the inquirer to appropriate sources of information about such services.

GARD views itself as working in partnership with the health-care community, always strongly recommending that patients seek the advice of their own health-care providers with any questions regarding their medical care. That collaborative approach has drawn praise from one nurse who turned to GARD for answers: “What I like best is how generous you are in your links to other organizations, which I think communicates concern for the person. As a nurse, I also like the advice encouraging people to consult a professional. I appreciate your response!”

In addition to furnishing health-care professionals with accurate, up-to-date information on genetic and rare diseases, GARD can also serve as a timesaving tool to help such professionals reinforce or expand upon genetic and rare disease information that they may have already provided to patients.

One patient who used GARD’s services sums it up in this way: “Thank you so much for your help and the additional resources that you’ve listed. So many times, sad news is shocking and we cannot or do not remember the advice the doctors gave us. I appreciate your time and effort.”